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ABSTRACT

Intended for use by the public as well as by medical professionals and related service agencies, the booklet presents genetic counseling as a means of providing information to deal with genetic disorders in general and mongolism (Down's syndrome) in particular. Characteristics of mongolism and possible emotional effects on the family of a mongoloid child are explained. Hereditary factors involved in mongolism are discussed. The birth defect is described to be either inherited or, more frequently, caused by a genetic accident (mutation) occurring at conception. Described are new genetic and genetic counseling procedures involved in preventing mongolism, such as amniocentesis and chromosome analysis. Answered are some specific questions concerning procedures used and recommendations often made in genetic counseling. The application of genetic counseling to the prevention of other birth defects is also discussed briefly. (KW)

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Genetic Counseling and Mongolism (Down's Syndrome)

Prediction
Detection
Prevention

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Preface

This booklet is designed for use by the public as well as by people in the medical profession and related service agencies. The terms "mongolism" and "Down's syndrome" are used interchangeably, but the term "mongoloid child" is used instead of the preferred medical term, "downic child," because of the laymen's greater familiarity with the former.

Genetic counseling is presented as an area which can provide information for dealing with hereditary disorders. The author does not, however, mean to infer that receiving information on genetics be necessarily followed by counseling.

The issue of institutionalization of mongoloids is presented because the author believes this to be an important one to parents. The modern trend seems to be away from institutionalization. In the past, however, some doctors have recommended such action even before the parents have had a chance to adjust to having a child who is afflicted with mongolism.

The author believes that this booklet will be of value to people in the medical profession; people in service agencies; anyone interested in birth defects and their elimination; and especially, to parents and relatives of children afflicted with mongolism or other birth defects. In writing the author hypothesized that mongolism can be predicted; its carriers identified; afflicted fetuses determined; and the defect itself eliminated.

The chromosome karyotypes that appear in this booklet were provided through the courtesy of Leonard Atkins, M. D., of the Massachusetts General Hospital. The author would also like to acknowledge Dr. William Goldman and Dr. Neal Anderson of Fitchburg State College and Lewis Holmes, M. D., of the Genetic Unit at Massachusetts General Hospital who were particularly helpful and cooperative. The following organizatons also aided in providing information that was used in the writing of this booklet:

National Association of Mental Health
National Association for Retarded Children
National Foundation — March of Dimes
National Institute of Neurological Diseases and Stroke

CHAPTER ONE

Introduction

The American Association on Mental Deficiency has described mental retardation as significant "subaverage general intellectual functioning which originates during the devlopmental period and is associated with impairment in adaptive behavior." Estimates have been made that 3% of the population of the United States (over 6 million people) are mentally retarded and should be identified as such before they are 15 years old.[1] The cost in support of schools, residential institutions, and specialized services for the mentally retarded costs the taxpayer over half a billion dollars each year. A sum even larger than this is borne by the families of the retarded. Mongolism, as well as many other defects, is characterized by mental retardation and physical limitations.

Modern medicine with its ever increasing knowledge and surgical improvements is now able to do much for victims of congenital heart defects, cleft palate and cleft lip, split spine or spina bifida, hydrocephalus, clubfoot, imperforate anus, genito-urinary defects, and many other birth defects. Whether the defect requires blood transfusions as with Rh factor babies, surgery, plastic surgery, prosthetic appliances, or special diets it seems that in practically all cases some resources of help are available.

Modern medicine can now also accurately predict the possibility of many defects occurring in specific family instances. In an ever increasing number of cases parents no longer have to accept the explanation that the birth of a defective child to their family was just one of those "unexplainable things." Many couples can now find out whether a birth defect in their family was in some way a hereditary one, and what the chances are of this defect being passed on.

If the defect was one that might be passed on to future children a test of the amniotic fluid surrounding a developing baby may tell whether or not the defect will be present in that child. If the test should show that the child would have the defect the parents may then wish to have a therapeutic abortion and try again later for a normal healthy child. Needless worry and hardship can be spared many families and many children can be spared the tragedy of birth defects.

Estimates have been made that one baby in every sixteen born in our country has a serious defect. To put this another way, everyday in the United States over 700 babies are born with defects serious enough to cause physical and mental handicaps to them and anguish to their families.[2]

With one child in sixteen having a serious birth defect

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it would be rare indeed to find a family tree that does not have some mark of imperfection in the past. So, if a person wants to blame somebody, he'll probably be able to find some excuse. That, however, is not the purpose of genetic counseling or this booklet. The prime consideration is trying to find out how such defects can be overcome or eliminated in the future.

Will the baby have this defect? This question is similar to the question, "Is the baby all right, Doctor?". The difference is that the question which the genetic counselor might answer can be answered before the child is born and often even before it is conceived.

Most parents of children with birth defects will not be satisfied until they have eliminated all possibilities that the child's retardation or handicap does not have some physical basis that will respond to medical treatment. Would it not seem reasonable then, that if they could, they would do as much for another child before it was born while there is still time to correct possible errors? This help before a child is born or even conceived can be given through genetic counseling.

This booklet gives an in-depth explanation of the applications of genetic tests and genetic counseling to the birth defect known as mongolism or Down's syndrome. Reference is also made to how genetic testing and genetic counseling can be applied to many other defects. It is designed to illustrate how the parents, brothers and sisters, or relatives of a defective child (in this case a child with mongolism) can determine the chance of their giving birth to another such child, and, if possible, prevent it.

The author can personally justify the need for a solution to the problem of mongolism because he has a sister who is afflicted with this defect. He is married and not too long ago his wife was with child. He wondered whether there was any increased chance of his wife giving birth to a child with mongolism. He sought answers to his questions and shares them along with other information in this booklet. Genetic testing and genetic counseling procedures allowed him to be assured that he would be the father of a girl who would not have mongolism, and he, quite proudly, is.

MONGOLISM (DOWN'S SYNDROME) IS A COMMON AND COSTLY BIRTH DEFECT CHARACTERIZED BY DISTINGUISHABLE PHYSICAL AND EMOTIONAL FEATURES AND MENTAL RETARDATION.

Dr. John Langdon-Down noted that the people afflicted with this defect had some features such as slightly slanted eyes

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that resembled the Oriental and so coined the name "mongolism." Because of this, individuals with this defect are said to have "mongolism" and are often referred to as "mongoloids." It really has nothing to do with the Orient and is only a way to classify a physical type.

The National Foundation — March of Dimes reports that mongolism occurs in about 1 in 600 births. In other words, thousands of infants with Down's syndrome are born in the United States each year. It appears in very intelligent as well as poorly educated families, rich and poor, in all races. With no known cure for mongolism help for this individual is limited. Families who decide to keep such a child at home are frequently troubled by the extra financial burden of needed services, and even with special training IQ usually does not improve to any great extent. Estimates of the cost of care for one mongoloid placed in an institution average about one-quarter of a million dollars for lifetime institutionalization. [3] Whatever the true figure is, the expense represents a serious burden to society.

What Are the Characteristics of Mongolism?

The mongoloid child is different from others in many ways. He has distinguishing physical, emotional, and intellectual features by which he can be identified. The following is a list of features, some of which the child will have and others of which he may have:

1 An extra chromosome, number 21, or its equivalent
2 A broad short skull
3 A round head
4 Slanting eyes
5 An unusual pattern in the iris of the eye, often with white specks
6 A short "saddle" nose
7 A large protruding tongue
8 A protuding lower lip
9 Shorter than usual height
10 Short and stubby hands
11 An unusual crease extending across each palm
12 Little fingers curving inward
13 Thick short feet
14 Tibial arches on each foot
15 A cheerful and affectionate disposition
16 Stubbornness
17 Mental retardation
18 Slow physical development

The mongoloid child has many handicaps. Mental retardation and deficiency are perhaps the most disheartening. Mongoloid children range intellectually from very severely retarded to near normal, but the majority of them achieve only between one-quarter and one-half of normal intelligence.[4] These children have an increased risk of having cataracts or crossed eyes, hernias, congenital heart defects, and a high susceptibility to respiratory disorders. Mongoloid children are also 20 times more likely than normal individuals to have leukemia.[5]

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What Is Life Like for a Mongoloid Individual?

The brain and body damage of Down's syndrome is probably done before the 8th week of pregnancy, and since there is no remedy, the life of the mongoloid child is relatively unproductive. He is sensitive to infection and prior to the discovery of antibiotics children so afflicted rarely lived past their 10th year.[6] Mongoloid children now live well beyond their 20th year and because of this the incidence of living mongoloids has increased more than threefold within the last generation. The child lags behind in physical development such as sitting, crawling, and walking and sometimes does not talk until 6 years of age or longer.

Placing a child in an institution or keeping a child at home both have positive and negative aspects. If the child is placed in an institution his future is educationally dim. In an institution, the mongoloid may, however, find himself with "others of his kind" whom he can relate with. If the child is kept at home he is usually more sociable and more productive. At home, because of slower physical development and retarded mental development, the child, however, frequently plays with brothers and sisters, youngsters much younger than himself, or no one at all. The child is often an object of family guilt or shame and not infrequently is aware of this.

Many communities provide special classes to which a mongoloid may be admitted and find himself in a trainable or educable setting. Some mongoloids progress enough to be involved in sheltered workshop situations and others may even be holding down part-time jobs with understanding employers.

Most individuals afflicted with Down's syndrome, if not placed in an institution, live with their families for the rest of their life. Marriage is usually out of the question and sterilization is more frequently being considered advisable. Male mongoloids have not been known to reproduce, but females afflicted with mongolism have been known to bear children. If a mongoloid female is impregnated by a normal male there is a 50% chance that the child will be mongoloid and a 50% chance that the child will be normal **(See page 12)**. Even if a mongoloid gave birth to a normal child the mother would probably not be able to adequately care for it.

What Is Life Like for the Family With a Mongoloid Child?

More than 6000 couples in the United States alone gave birth to a mongoloid child last year. One of their first decisions, after the shock of this event had passed or even before, was whether to keep the child at home or to have the child placed in an institution. Placement is usually done

shortly after birth. However, parents may decide to keep the child at home and later discover that they cannot provide the time and care needed by such a child. The child may be placed in a state institution, where space is available, or if the parents are more financially able the child may be placed in a private institution or special school.

If the mongoloid child is placed in an institution the family contact becomes limited. Parents usually make visits to their child and sometimes take him or her home for periodic vacations. Most of the negative social stigma of having a defective child is avoided when the child is away from the home, but parents and children must still tell how many people are in their family and account for the mongoloid individual who is missing or only visits a few times a year.

When the mongoloid child is kept at home the family needs understanding and acceptance of this individual. Parents must understand the limitations of their child and yet still be willing and able to devote long hours to care and training of the child. The family's activities are frequently hampered by the mongoloid child and parents may fear leaving the child unattended or with someone else. Worry may also exist over whether the child will hurt himself or other children.

Some parents feel ashamed or guilty because they fostered a defective child. Other parents may ignore the child or even pretend it was never born. Still others can accept the birth of a mongoloid child and make good adjustments for handling the situation.

Brothers and sisters must usually help with the care of the mongoloid child. They watch after the child and play with him or her when other youngsters won't. They may get upset by degrading remarks made by others and sometimes get into a fight protecting the family image or the retarded mongoloid.

The parents must decide what course of action will be best for the mongoloid child and for the rest of the family. The author wishes to note, however, that having a mongoloid child at home need not always be considered negative. His observations have shown that families who do have a mongoloid living at home tend to be more open, more concerned with others, and more accepting of life.

The family, as it has been shown, must in some way or other deal with the problem of having a mongoloid child. Two other very serious questions face this family: Why did they have a mongoloid child? What are the chances that they will have another mongoloid child? In the next chapter the author will provide information that will help answer these questions.

CHAPTER TWO

MONGOLISM IS A BIRTH DEFECT WHICH IS EITHER INHERITED OR, AS MORE FREQUENTLY HAPPENS, CAUSED ENVIRONMENTALLY, USUALLY THE RESULT OF A GENETIC ACCIDENT (MUTATION) AT CONCEPTION.

Hereditary material from each of our parents made us develop into the human beings we are, with certain racial and familial traits. Tiny chromosomes are the particles in the cells which carry the "blueprint" for each individual in still smaller particles of hereditary matter called genes.

A normal child begins as a fertilized egg with 46 chromosomes which results from the union of an egg and a sperm which each contribute 23 chromosomes. A victim of mongolism has an extra chromosome or its equivalent, resulting in 47 chromosomes instead of the normal 46 or 46 chromosomes with extra material attached to one chromosome in what is called a "translocation". The first type (47 chromosomes) is usually the result of a genetic accident and is rarely hereditary. The translocation type (46+ chromosomes) is hereditary. Another type of mongolism called mosaicism also exists but it is very rare and is not hereditary.

How Does a Child Get the Extra Chromosome or Extra Chromosome Material That Makes the Child a Mongoloid?

The mother of a normal child contributes 23 chromosomes in her egg and the father contributes 23 chromosomes in his sperm. This means that a developing fetus starts from a fertilized egg with 23 pairs of chromosomes. Each pair of chromosomes is responsible for transmitting certain hereditary information. The extra chromosome or extra chromosome material found in mongoloids is associated with the 21st pair of chromosomes.

Each normal parent has 46 chromosomes in his or her cells and yet contributes only 23 chromosomes to the developing child. How is this done? It is done during the process by which the sperm and egg are formed. The 23 pairs of chromosomes in one cell divide into two cells each having 23 single chromosomes. These cells with only 23 chromosomes are the reproductive cells called the egg and the sperm cells.

The most common type of mongolism, **trisomy 21,** is caused because the 21st pair of chromosomes failed to divide and separate. This caused one sperm or egg cell to have 22 chromosomes and another to have 24 chromosomes. If for

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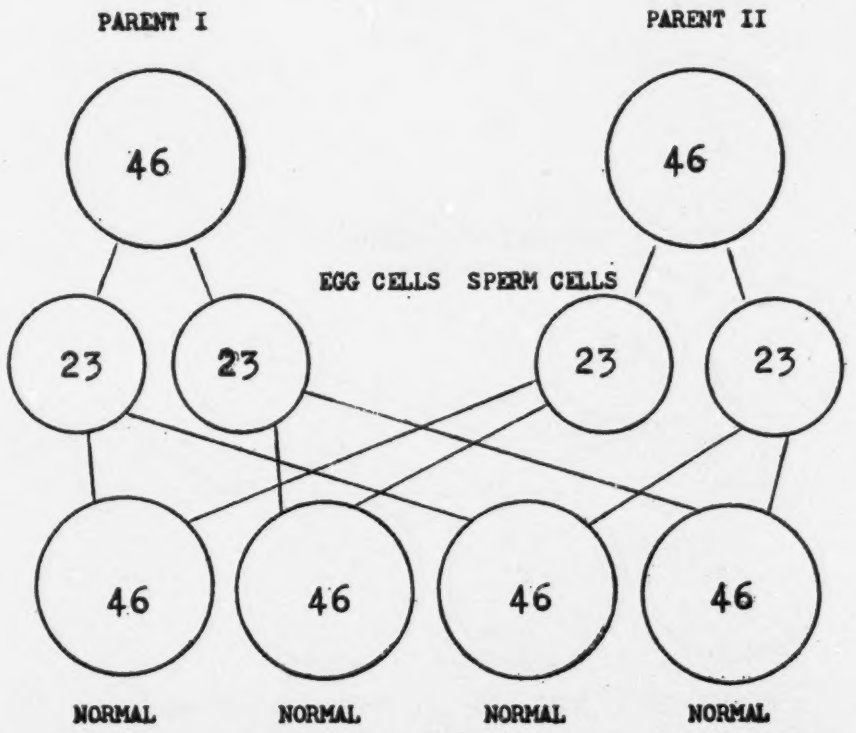
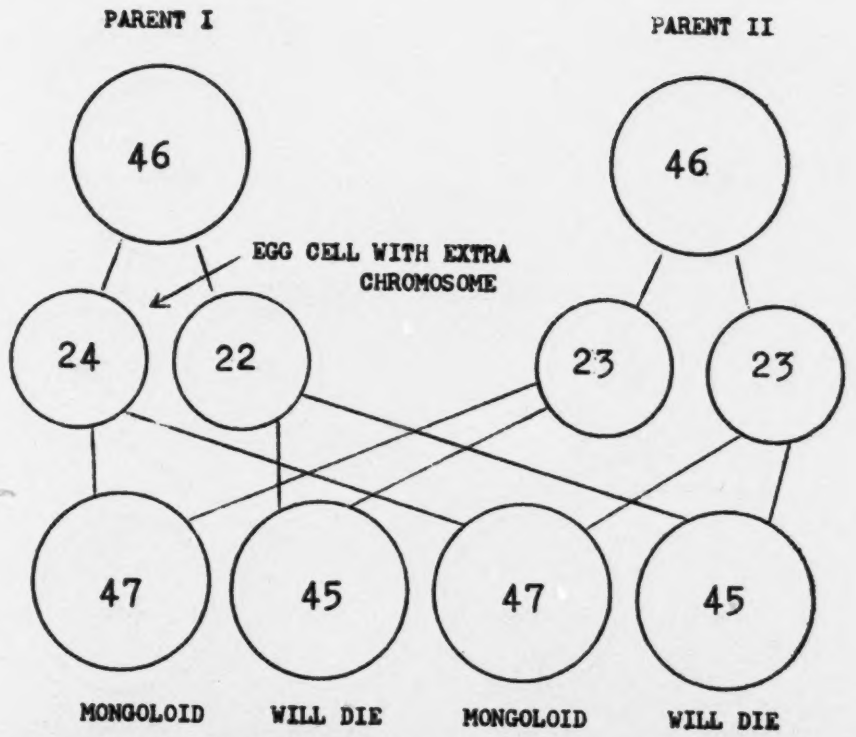


The cells of each normal parent have 46 chromosomes. When these cells divide they separate into two cells (eggs or sperm) with 23 chromosomes each. When an egg cell with 23 chromosomes is fertilized by a sperm cell with 23 chromosomes then a normal child with 46 chromosomes should develop.

instance an egg cell with 24 chromosomes is fertilized by a normal sperm cell with 23 chromosomes then the developing fetus would have 47 chromosomes. This child with 47 chromosomes would then be classified as a mongoloid. A child with 45 chromosomes (a 22 and 23 combination) does not develop because this chromosome makeup is usually not sufficient to sustain human life.

The **translocation** type of mongolism which is hereditary may be passed on by a carrier. The carrier has the normal amount of chromosome material but one of the #21 chromosomes is attached to another chromosome. When the carrier's cells divide there is a 25% chance that the chromosome with the #21 chromosome attached may go to form the same sperm or egg cell as the other #21 chromosome. If this egg or sperm then goes into making a fertilized egg a mongoloid child will result. The carrier may also produce a normal child or another carrier.



If one of the pairs of chromosomes (the 21st pair) in one parent fails to divide and separate this will cause an egg or sperm cell with an extra chromosome. When the sperm or egg cell with the extra chromosome goes into making a fertilized egg, a child with 47 chromosomes (mongolism) will result. The fertilized eggs with 45 chromosomes will die because this arrangement cannot support life.

What Hereditary Factors Are Involved In Mongolism?

Most cases of mongolism are the result of a genetic accident and are not hereditary. For this reason most parents of a mongoloid child have little increased risk of having another mongoloid.

The translocation type of mongolism is hereditary. Although it appears in less than 5 per cent of the cases of mongolism, these cases might have been predicted and prevented. Translocations of chromosomes #21 have been found in mothers, fathers, and even grandparents of a person with the translocation type of mongolism. Future children of such individuals will also be subject to mongolism. There is evidence, however, to suggest that many translocation mongoloids are borne of parents neither of whom are translocation carriers.[1]

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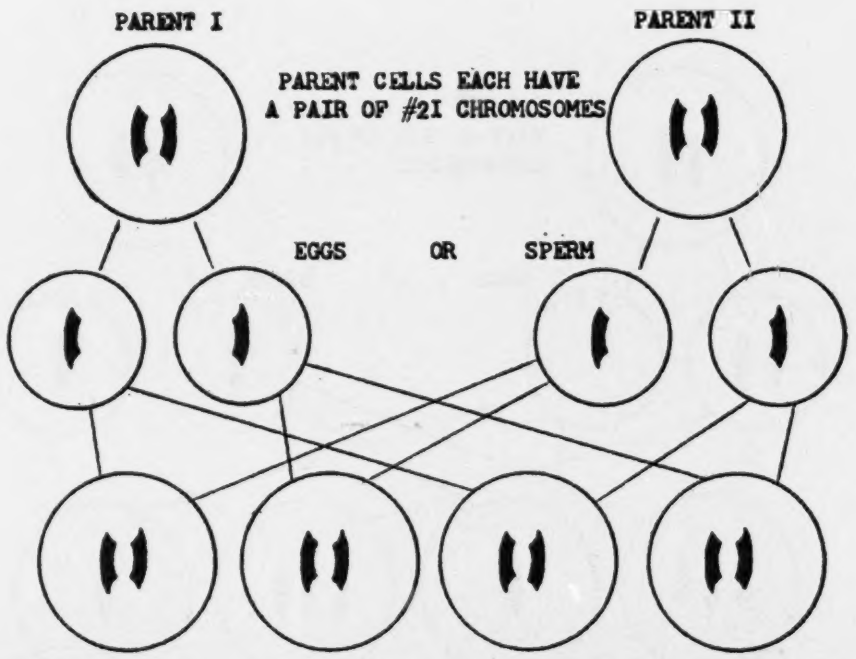


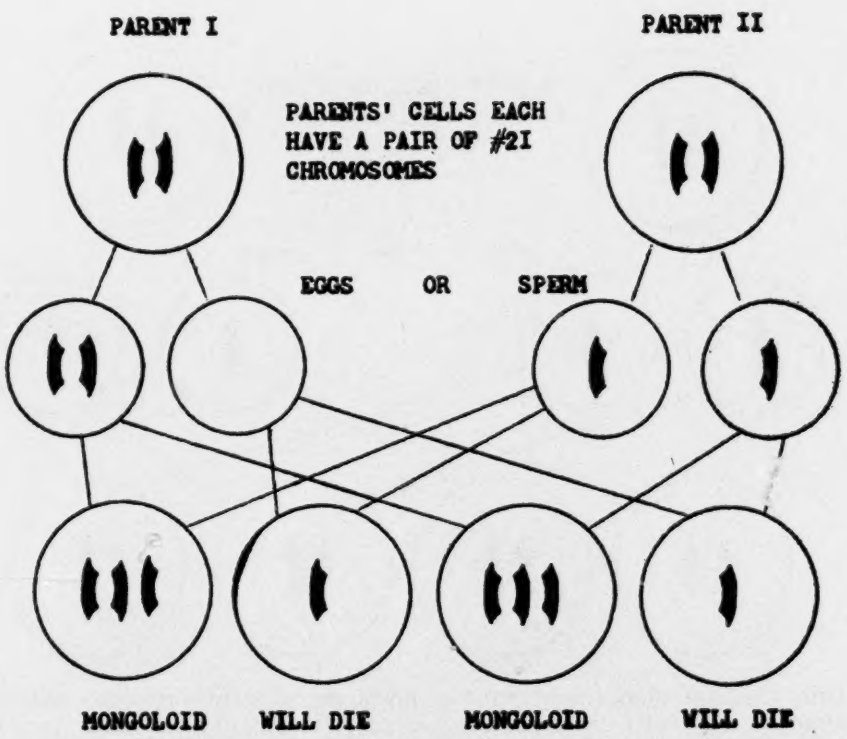
This diagram shows how normal pairs of #21 chromosomes divide, separate and then come together during fertilization in order to produce a normal child. All of the 23 pairs of chromosomes undergo similar processes. When a pair of #21 chromosomes fails to separate (nondisjunction) then there is a good chance that mongolism will result.

Is the Mother's Age Important in the Occurrence of Mongolism?

Yes. Very young mothers or very old mothers run a greater risk of having a mongoloid child. The National Foundation — March of Dimes reports women 25 years of age have about 1 chance in 2000 of producing a mongoloid child, while for women of 45 years of age the chance is increased to about 1 in 50. In general, pregnant women beyond the age of 40 have a 2 per cent to 3 per cent chance of giving birth to a baby with mongolism or some other chromosome disorder.[2]

In the human female, the sex cells which eventually become the eggs are present before the girl is born. The older the woman is, the older her sex cells are. The older sex cell is more likely to make a mistake when it divides because it has been exposed to more harmful environmental agents.

The author has been unable to find reasons for increased chances of mongolism in women who are very young, but suspects that this may be associated with failure of eggs to be



Both of these parents are normal, but the 21st pair of chromosomes in parent I has failed to separate in the formation of the eggs or sperm. When this egg or sperm comes together to form a fertilized egg, the fetus will become a mongoloid because of the extra chromosome.

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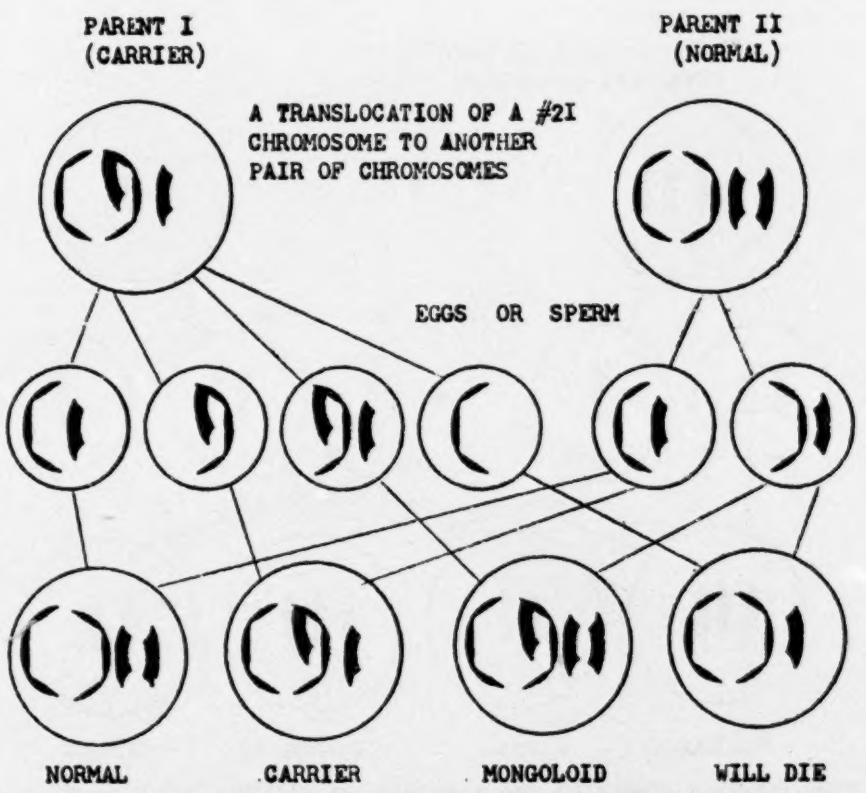
fully matured and developed. Mongoloid babies would also tend to be born to very young mothers who are translocation carriers.

Is the Father's Age Important in the Occurrence of Mongolism?

No, not really, unless one considers the fact that older men tend to be married to women who are also older. The male sperm, unlike the females' eggs, are constantly reproduced and so are not exposed to as many harmful environmental agents.

What Are Some Harmful Environmental Agents Which Might Increase the Chances of Mongolism?

X-rays and radioactive fallout have both been suspect as harmful environmental agents. Frequent exposure to large amounts of X-rays over extended periods of time can produce



Parent I is a carrier of translocation type mongolism. The carrier has the proper amount of chromosome material but in the wrong place (translocated to another chromosome). When the cell divides and separates the #21 chromosome material will come together in fertilization so as to produce mongolism in 1 of 4 cases. Since one of the four will die the carrier will produce mongolism in 1 of 3 children.

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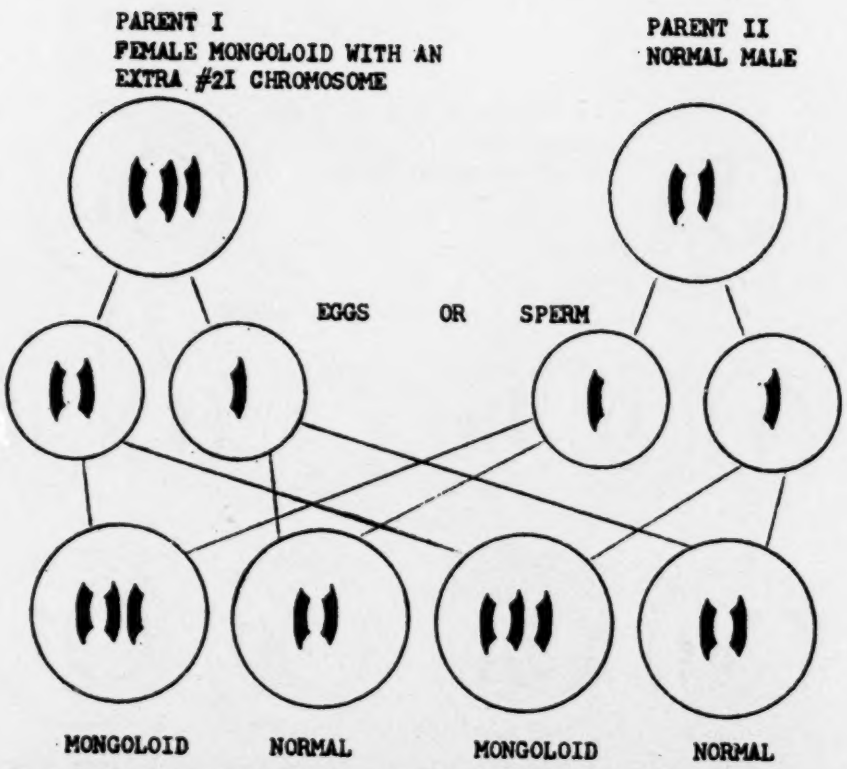
the mistake in sex cell division which causes mongolism.[3] This does not, however, mean that the use of X-rays by qualified professionals is harmful. The bacteria or viruses of such diseases as infectious hepatitis, German measles, and even Asian flu may cause damage to the developing infant. Certain drugs such as LSD, especially if taken near the time of ovulation or early in pregnancy, may be harmful. Anti-cancer drugs and certain hormones are also known to cause defects.

Recent information suggests that German measles has been blamed for causing more damage than it actually does. German measles probably does cause cataracts, mental retardation, deafness, small head, and heart defects, but whether or not it is serious enough to cause a chromosome disorder such as mongolism is highly doubtful.



This diagram illustrates the possibility of children being borne of a female with mongolism and a normal male. Very few mongoloids have borne children, but as is shown here, the possibility exists, because of the laws of genetics, that they could produce normal babies as well as other mongoloids. A study by Lejeune (1964) of 11 female mongoloids who were pregnant supported the fact that half of the offspring of such mothers could be expected to be be affected and half could be expected to be normal.[4]

What Chance Does a Couple Which Has Produced One Mongoloid Child Have of Producing Another Mongoloid Child?

Mongolism occurs in about 1 in 600 births, and over 90 per cent of the cases of mongolism are caused by a genetic accident which is not hereditary. For this reason most families with one mongoloid child will not have another.

If, however, a family has a mongoloid child with a translocation type of mongolism this would tend to indicate that one of the parents may be a carrier of this type of mongolism which is hereditary. In order to find out whether the mongolism is the hereditary type or not, a chromosome study may be done. If chromosome studies show that the child has the translocation type of mongolism and a parent is a carrier of

mongolism, then for his family the chances of having another mongoloid child are about 1 in 3 births. Chromosome studies are discussed later in this booklet (see page 15).

What Chance Do Normal Brothers and Sisters of a Mongoloid Child Have of Producing Another Mongoloid Child?

If the mongoloid child was produced by a genetic accident, which is not hereditary, then normal brothers and sisters would have about the same chance as anyone else of producing a mongoloid child. This is particularly true if their mother was much beyond the age of 35 when the mongoloid child was born.

If the mongoloid child was produced by the hereditary type of mongolism (translocation of chromosome #21) the normal brothers and sisters would have a 50 per cent chance of being a carrier of mongolism. One in three of a carrier's children would be mongoloid, one normal and one normal but a carrier.

A chromosome study may be done for a normal brother or sister of a mongoloid child to find out whether he or she is a carrier. New genetic testing procedures discussed later in the booklet can then be used to insure that the carrier's children will not have mongolism (See amniocentesis, page 15).

CHAPTER THREE

NEW GENETIC AND GENETIC COUNSELING PROCEDURES ARE INVOLVED IN PREVENTING MONGOLISM AND OTHER BIRTH DEFECTS.

Genetic counseling developed out of an in-depth investigation of how chromosomes transmit genetic information and is mainly concerned with keeping hereditary disorders from being transmitted. In its early stages genetic counseling was used just to predict the chances of a person passing on a hereditary defect, but it has now become a more exact diagnostic tool.

Genetics can help many couples who dare not have a child or another child for fear of passing on an inherited disease or defect such as mongolism. Genetic specialists try to assure parents that their children will not be afflicted with a hereditary defect and aim at employing a safe and accurate method by which any pregnant woman can determine whether her baby will suffer from genetic abnormalities.

Early identification of a genetic defect can often mean that a resulting disorder can be treated satisfactorily. For example, PKU, galactosemia, congenital hypothyroidism, and many other inborn errors of metabolism can be beneficially

treated by such procedures as the early administering of special diets.

What Does Genetic Counseling Involve?

Genetic counseling, quite simply stated, involves using genetic and medical tests to find out whether someone has a hereditary defect or could be a carrier of a hereditary defect, and then counseling this person as to risks to future children and diagnostic and preventive measures that are available. Counseling may be conducted by a team or by a doctor who has already conferred with other medical people. Many geneticists state the statistical risk figures and present different tests and procedures that may be used, and abstain from suggesting any concrete measures. The person or couple being counseled must decide what action, if any, will be taken.

The first step in genetic counseling may involve reviewing the information of a referring physician, if there was one. After this, the couple will be questioned about their family medical history, preferably going back three generations. Questions may be about recurring diseases, incidence of miscarriage or still birth, whether the couple was related before marriage, how old relatives were when they died, and what the causes were. From all this information a diagram of the family's medical history called a "family pedigree" will be made. It can be used to see whether or not the defect may have been passed on from previous generations and can help in predicting the future.

The abnormal child will be examined, if possible, and the geneticist may want to do chromosome, enzyme, or other studies of the blood, urine, or skin cells of the couple in order to make an accurate diagnosis. If the disease or defect is suspected to be a hereditary one, affected brothers and sisters of the couple may also be examined.

There are many other tests and techniques that may be used and the following questions in this section refer to just a very few. Chromosome analysis and amniocentesis are given special attention because they are recent and have produced great beneficial advances.

What Is Intrauterine Diagnosis?

Intrauterine diagnosis involves examining the uterus of a woman and looking for problems which might exist. In many cases it involves evaluating the condition of a developing baby in a woman who is pregnant. Amniocentesis, which is discussed later in this chapter, is a form of intrauterine diagnosis, and it can be used in some cases to tell whether or not a developing baby will or will not have a hereditary defect if it is born.

What Is Chromosome Analysis?

As was stated earlier, chromosomes are units in the cells which carry hereditary material. Chromosome analysis involves the study of a person's chromosomes to determine whether or not the person suffers from a chromosome disorder or is a carrier of a chromosome disorder.

In order to study the chromosomes, cells taken from a blood sample or from a sample of amniotic fluid by amniocentesis are cultured in the laboratory. When the chromosomes in the cells have doubled during cell division the chromosomes are stained with a chemical and placed under a microscope where they are photographed. The photograph is enlarged and the individual chromosomes are cut out. The chromosomes are then matched by size and shape into pairs and transferred to what is called a "karyotype." A genetic counselor, by analyzing the karyotype, may then tell whether or not there is some kind of chromosome difficulty.

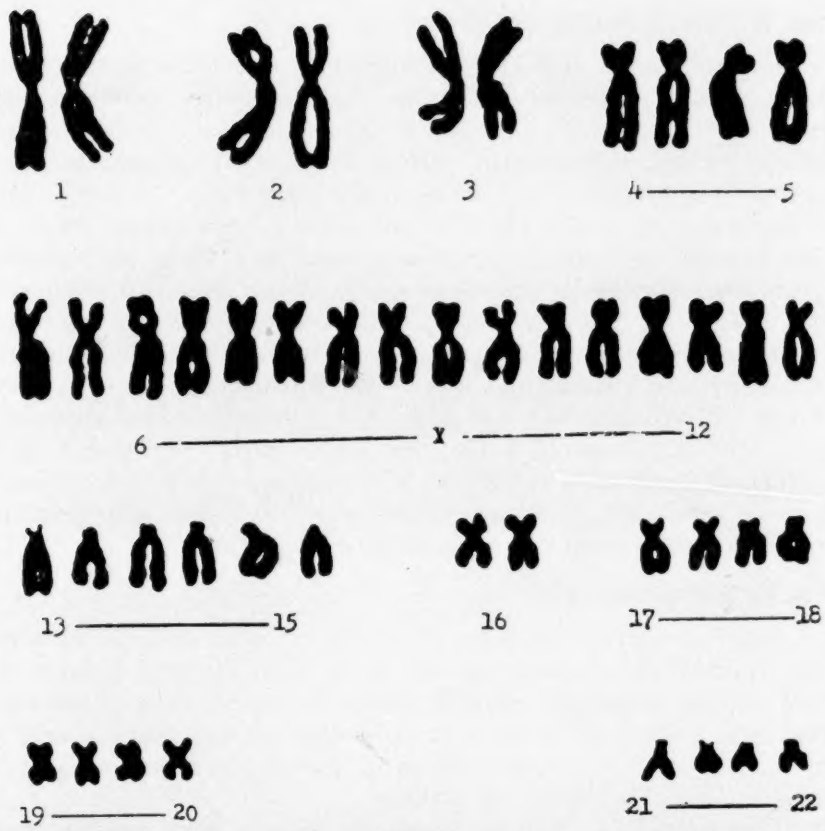
What Is Amniocentesis?

Amniocentesis involves obtaining a small sample of amniotic fluid which surrounds the fetus (developing baby) so that it can be examined. Examination of the amniotic fluid can detect mongolism, all gross chromosomal abnormalities, and a growing number of inborn errors of metabolism; all of which could produce a defective child.

The amniotic fluid is obtained with a thin needle that enters the amniotic sac that surrounds the baby through the abdomen (stomach) of the mother. A technique called "ultrasonic placental localization" is sometimes used to locate the position of the fetus before amniocentesis is performed, but this is not usually needed. The actual amniocentesis procedure takes only a few minutes, is not painful, requires no anesthesia although a local anesthetic may be used, and leaves only a small pin prick on the mother's stomach. The procedure is usually done in a doctor's office and the woman is allowed to go home afterwards. Occasionally, a good sample is not obtained on the first try and a retap may be scheduled at another time.

The optimum time to have an amniocentesis test performed seems to be between the 14th and 18th week of pregnancy.[1] This is long enough for sufficient amniotic fluid to accumulate and early enough for a safe therapeutic abortion, if the results should cause the mother to choose one. Laboratory tests on the amniotic fluid may take from two to three weeks, and it is usually best to have an abortion performed by the 20th week of pregnancy.

The decision to have a therapeutic abortion, if the suspected disorder is found in the developing baby, should be

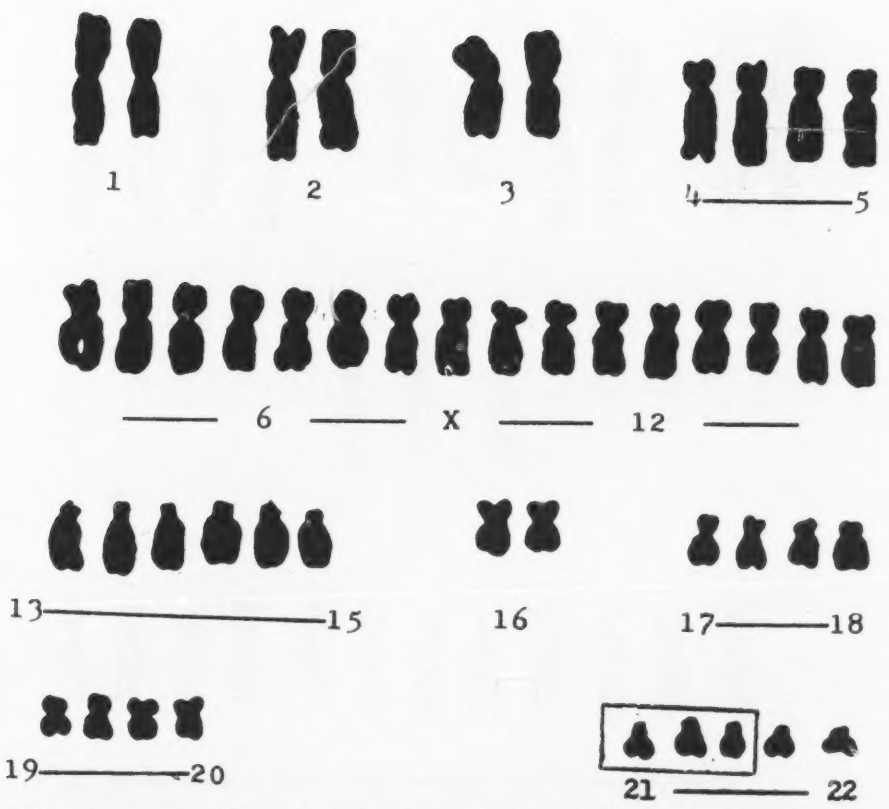


This is a chromosome karyotype which shows the 23 pairs of chromosomes of a normal female, numbered 1 through 6, X, and 7 through 22. The two X chromosomes indicate that the subject is female.

made before the amniocentesis is performed. There is a small risk to the mother and fetus, and because of this a genetic counselor will usually only recommend or do amniocentesis when there is reason to suspect a defect and the couple is prepared to act upon the results.

This intrauterine technique is frequently done to detect the presence of Down's syndrome which is the most common chromosomal disorder. Amniocentesis can also be used to detect other chromosome disorders such as double-Y, Kleinfelter's syndrome and Turner's syndrome. Amniocentesis can determine the sex of the fetus when dealing with recessive X-linked disorders such as hemophilia and muscular dystrophy, and has other uses, some of which are listed in chapter five.

The physician will discuss the pros and cons of amniocentesis with the couple seeking help or information. In the course of this discussion he will impress upon them that although testing through amniocentesis is almost 100 per cent



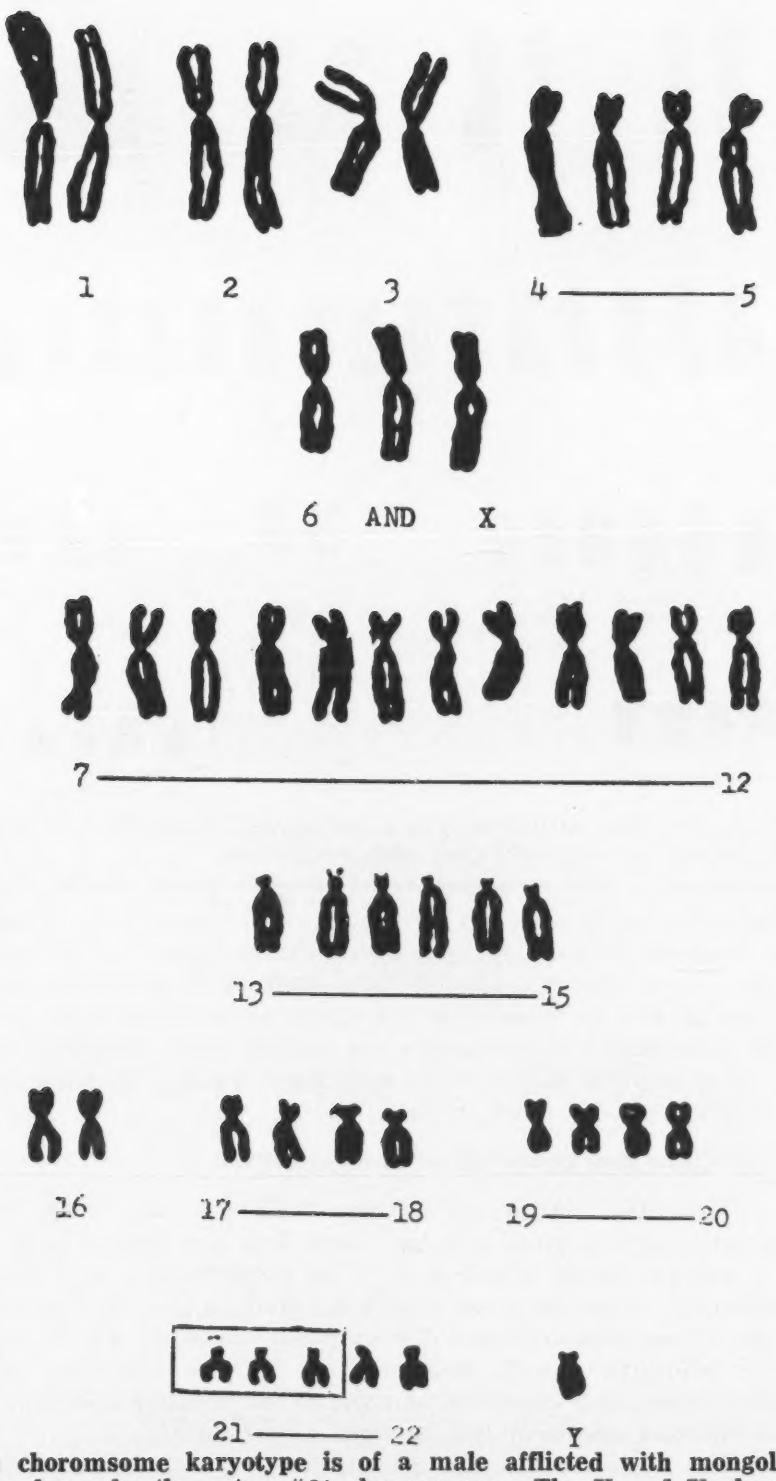
This chromosome karyotype is of a female with the extra #21 chromosome which indicates affliction with mongolism.

effective in detecting the presence of the particular disorder being tested, it does not guarantee that a normal child will be born. Tests on the amniotic fluid such as chromosome analysis can usually be done only for one type of disorder at a time. This is because the geneticists are usually only looking for one disorder and the laboratories only have a small amount of the amniotic fluid to work with.

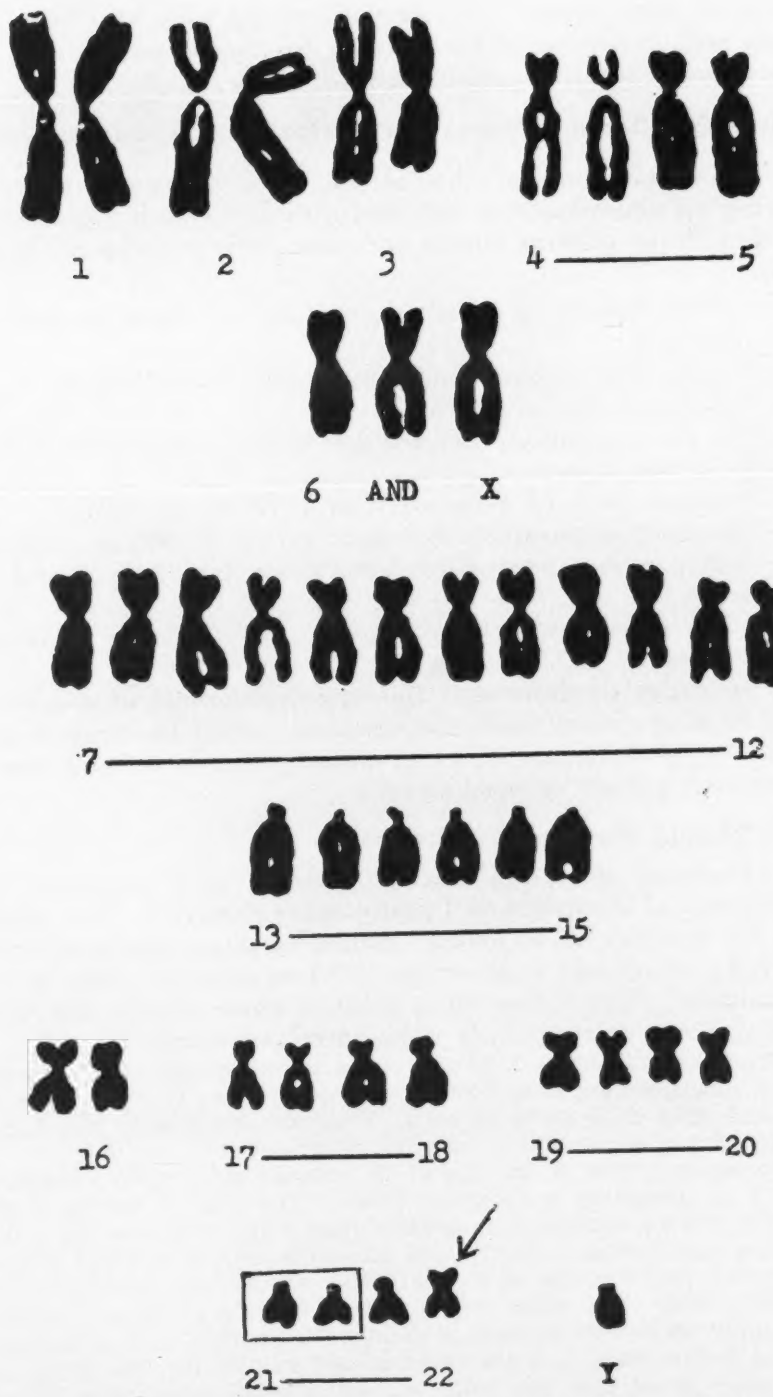
What Risks Are Involved in Amniocentesis?

The risks involved in amniocentesis are very small, estimated at less than one per cent, but the author feels that they deserve to be mentioned. The greatest risk, and the one that seems to be the most clearly verified, is that the procedure might cause miscarriage. Other risks, some of which studies have failed to clearly substantiate, include bleeding, blood sensitization, and infection as risks to the mother and puncture and induced malformation as risks to the child.[2]

The values and risks of amniocentesis can be better described for a particular couple's situation by a genetic coun-



This choromsome karyotype is of a male afflicted with mongolism as is shown by the extra #21 chromosome. The X and Y chromosomes indicate that this subject is a male.



This chromosome karyotype is of a male mongoloid who has extra #21 chromosome material translocated to a chromosome in the #22 chromsome pair. The translocation indicates that the mongolism may be hereditary.

selor or obstetrician. The answers to the following two questions will, however, show reasons for which amniocentesis is done and when it is considered advisable.

What Are Some Reasons for Performing Amniocentesis?

The following is a list of the most common reasons for having an amniocentesis test performed.[3] This list is arranged so that those reasons which are most widely accepted appear first.

1—Family history of gene-transmitted or chromosomal abnormality.
2—Parent with balanced chromosomal translocation.
3—Advanced age of mother.
4—Sex determination, i.e. for predicting sex-linked defects.
5—Rh blood factor.
6—Previous birth of a retarded or malformed child.
7—History of exposure to mutagen(s), i.e. X-rays or radiation.
8—Poor previous pregnancy history, i.e. miscarriage and still births.
9—Viral infection first trimester, i.e. Rubella (German measles).

In order to determine the appropriateness of amniocentesis to a particular case consultation should be made with a doctor or obstetrician who can either give advice or make referral to a genetic counseling unit.

Who Should Have an Amniocentesis Test?

Prenatal detection can provide a firm diagnosis for chromosomal disorders and many other disorders. The patient and the obstetrician or genetic counselor must, however, weigh the risks of having a defective child against the risks of amniocentesis. The following is a list of three groups for which amniocentesis is frequently considered advisable:

1—When one parent is a carrier of a chromosomal rearrangement (i.e. translocation type Down's syndrome) there is a high risk that a defective child could be born. Pregnant mongoloids also have a high risk of producing a defective child.
2—Women pregnant at the age of 40 years or older have a moderate risk of producing a defective child. (The risk of having a child with Down's syndrome is greater than 1 per cent and the risk of other chromosomal aberrations also increases in this age group).[4]
3—Women past the age of 35 years and women who have previously given birth to a child with trisomic Down's syndrome which is usually not hereditary have a greater than normal risk of producing a defectve child. (Amniocentesis test results for this group will usually show that the child will be normal but performing amniocentesis may be justifed on the basis of assuring a family that this child will not have a chromosomal disorder such as Down's syndrome.)

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What Do the Doctors Have to Say About Genetic Counseling and Mongolism?

The following information was gathered through a questionnaire on Down's syndrome (mongolism) and genetic counseling which was sent to 60 obstetricians selected at random through the Yellow Pages of Massachusetts' telephone directories. This information is based on the 41 obstetricians who answered the questionnaires.

1. Have you ever delivered a baby with birth defects?
   Yes **41** No **0**
2. Have any of your patients ever given birth to a child afflicted with Down's syndrome?
   Yes **37** No **4**
3. Do you presently have any genetic problems in your practice?
   Yes **17** No **22**
4. Have you ever given or recommended genetic counseling to any of your patients?
   Yes **38** No **3**
5. Do you think that parents who have one mongoloid child should get genetic counseling, even though most cases of Down's syndrome are not passed down by heredity?
   Yes **38** No **3**
6. Do you think that a normal sibling (brother or sister) of a mongoloid child should seek some kind of genetic counseling when he or she plans to have children?
   Yes **17**
   Yes **20,** if there is reason to believe the mongolism may be hereditary.
   No **6**

*7. Have you ever recommended to one of your patients that she have an amniocentesis test?
   Yes **36** No **5**
8. Have any of your patients ever had an amniocentesis test?
   Yes **37** No **4**
9. How familiar are you with genetic counseling services to which you could refer a patient in need of such services?
   31 I know where I could refer a patient
   7 I would have to investigate genetic counseling services in my area before referring a patient
   2 I have little present knowledge of such services

*10. What is your opinion of the usefulness of amniocentesis?
   0 Little proven value at this time
   15 Useful for only a very small minority with definite hereditary problems.
   12 Useful for patients who question the possibility of passing on a hereditary defect

26 Useful for patients who question the possibility of of passing on a hereditary defect, or question dangers due to exposure to Rubella or increased maternal age

11. What is your estimate of the risk involved in having an amniocentesis test performed?
    - **13** Negligible
    - **15** Very little, considering future damage it can help avoid
    - **11** Moderate, the patient should be well aware of the pros and cons involved
    - **2** High, to be considered seriously and with caution
    - **0** Not sure
12. Dr. Hans Zellweger, of the University of Iowa's Hospitals has advised performing amniocentesis in all pregnant women more than 40 years old, and in all others with a high risk of carrying an abnormal fetus. Do you think think that this will come to be a widely accepted practice within the next 10 years?

    Yes **28** No **11**

* To question #7 six doctors stated that they had recommended an amniocentesis to their patients, but only because of Rh blood sensitization problems.

* To question #10 a few doctors (3) questioned the use of amniocentesis because of exposure to German measles. As was stated earlier in this booklet German measles is probably not serious enough to produce chromosome damage.

Numerical discrepancy is accounted for by a few obstetricians who either neglected to answer a specific question or chose more than one answer to a certain question.

Who Can Have and Who Should Have Genetic Counseling?

Anyone can consult an obstetrician or genetic counselor about questions referring to a family disorder which may be hereditary. Actual testing procedures would probably only be recommended if the counselor felt that there was a greater than normal risk of producing an abnormal child. Testing procedures may, however, also be advised, even when it is felt that normal risks exist, if it would relieve great anxieties in worried parents.

Genetic specialists are visited most often by couples who have already had a defective child or by couples whose family histories include birth defects. Women who have been exposed to mutagenic (gene changing) risks such as German measles, overdoses of radiation or X-rays, and chromosome damaging chemicals or drugs may also seek genetic counseling. Anyone who has reason to believe that he or she may pass on or produce a defect in a newborn child should consult a genetic

counselor. The counseling that this person or couple may receive could relieve worries now and serious guilt feelings later.

CHAPTER FOUR

THERE ARE SPECIFIC PROCEDURES THAT GENETIC COUNSELING OFFERS WHICH CAN BE USED TO PREVENT THE BIRTH OF BABIES AFFLICTED WITH MONGOLISM.

The birth of mongoloid babies because of hereditary factors can be eliminated. Chromosome analysis of mongoloids at birth would indicate in each case whether or not the mongolism was of the hereditary type. If the mongolism was of the hereditary type chromosome analysis of the parents would indicate if one of the parents was a carrier. If one of the parents were found to be a carrier of mongolism the chances of this couple producing another mongoloid child would be high (1 in 3). This couple could still have more children, however, without being afraid of having another mongoloid child. Amniocentesis could detect mongolism in a developing baby and a therapeutic abortion could eliminate the birth of the child who would be defective because of mongolism. The couple would then be free to try again for a normal child.

The birth of babies with mongolism which is not hereditary can also be eliminated in many cases. The use of amniocentesis in older women who are pregnant can show whether or not the developing baby would have mongolism. Here again, a therapeutic abortion could eliminate the birth of a child who would be afflicted with this defect.

The following questions in this section relate to genetic counseling and mongolism in a more detailed manner. The author hopes that the questions will be ones that a couple seeking genetic counseling might have and hopes that the answers will be informative.

What Can a Couple Do to Avoid Having a Mongoloid Child?

The two most important things that can be done to avoid having a mongoloid child are to have a healthy pregnancy and to make use of new medical knowledge, tests and techniques which are available.

If a couple has already had one mongoloid child a chromosome study of the child should be done. The chromosome study can confirm the diagnosis of mongolism and indicate whether the mongolism may be hereditary. In cases where the mongolism is the result of translocation (the hereditary type of mongolism) the mother may often have a chromosome abnormality and in some cases the father has

had a chromosome translocation.[1] Chromosome studies of the parents are recommended because of the increased risk that either of these parents might subject future children to Down's syndrome. The chromosome analysis will tell whether or not either of the parents is a carrier of mongolism. If one of the couple is a carrier of mongolism then other children of these parents should also have a chromosome study done.

What Can a Normal Brother or Sister of a Mongoloid Child Do to Avoid Producing Another Mongoloid Child?

The normal brother or sister of a mongoloid child can find out if the mongolism may be hereditary if a chromosome study is done on the child with mongolism. This information will help all members of the family. If for some reason, however, a chromosome study cannot be done on the affected child, a normal brother or sister could still find out if he or she was a carrier by having a chromosome analysis done on himself or herself.

What Factors Are Involved in Any Healthy Pregnancy?

Many factors contribute to a healthy pregnancy. The most helpful information on pregnancy can probably be obtained from your family doctor or obstetrician. The following, however, is a list of factors that should be considered in relation to any pregnancy.

1—It is dangerous to be married to a close relative because errors in heredity may be compounded.
2—If your husband has been exposed to radiation accidents pregnancy should be postponed until it is determined safe by a doctor.
3—Let your doctor know as soon as you think you are pregnant.
4—Make sure your doctor is informed of your health history, your blood type and your husband's blood type, and the birth defects history of any relative.
5—Only take medicine or drugs, including aspirin, that are prescribed by your doctor who knows you are pregnant.
6—Make sure any and every doctor (including your dentist) who is treating you for anything knows you are pregnant.
7—Avoid stomach, low back and pelvic X-rays except in an emergency.
8—Use a good diet to feed yourself (and your baby).
9—Avoid exposure to any infectious or contagious diseases, especially German measles.
10—Avoid heavy smoking because it may affect the baby's birth weight which would be very important if the baby is born premature.

X . 29

What Tests or Techniques Can Be Used for a Person or Couple Who Questions the Possibility of Producing a Mongoloid Child?

A family's medical history can be used by a genetic counselor to help determine if mongolism in this family is hereditary. Chromosome analysis from a blood sample can be used to tell if someone has mongolism or if someone is a carrier of mongolism. Amniocentesis and chromosome analysis can be used to tell if a fetus in a pregnant women is mongoloid and a therapeutic abortion can be used to prevent the birth of a mongoloid child.

What Will Happen if Amniocentesis Shows That the Baby Will Be Mongoloid?

A therapeutic abortion is advised. Parents should not have an amniocentesis test performed unless they are prepared to act upon the results.

What Will Happen if Someone Is Found to Be a Carrier of Mongolism?

If a carrier of mongolism has children and takes no preventive measures against mongolism the chances are that one in three of the carrier's children will be mongoloid, one will be a carrier, and one will be normal. What this person and his or her spouse decides to do with this knowledge is up to them.

If this couple decides to have more children together, amniocentesis can be used to identify fetuses who would be affected with mongolism, and these could be therapeutically aborted. A normal fetus, one not affected with mongolism, would be allowed to continue in development and be born.

Since there is a small risk involved in amniocentesis, some couples, especially those who are older and already have other normal children, may consider some birth control method more suitable. In other cases where the couple does not wish to have an amniocentesis test performed sterilization, artificial insemination, or adoption may even be considered. Sterilization, if considered, should be done only on the individual who is the carrier. Adoption could be considered if the woman was the carrier or if the couple didn't wish to have any more of their own children by using amniocentesis.

Does a Couple Have to Wait Until the Woman Becomes Pregnant Before They Can Find Out if Their Child Will Have Mongolism?

Yes, amniocentesis can only be used to detect mongolism

in a fetus of a pregnant woman. Before a woman becomes pregnant a genetic counselor can only present a couple with their risks or chances of producing a mongoloid child.

Can a Chromosome Study Be Done for a Man or Woman Before He or She Is Married?

Yes. A chromosome analysis can be done for either a man or woman and it can be done before they are married. The chromosome analysis, in this case, would be done from a blood sample taken from the individual.

Is a Therapeutic Abortion Difficult to Obtain?

Abortion laws vary from state to state, but an abortion should not be difficult to obtain where one is needed for medical reasons which are in the best interests of both the child and the mother. Anti-abortion laws are based on the premise of protecting a yet unborn baby. By similar reasoning a therapeutic abortion is performed also to protect an unborn baby. Carrying a fetus afflicted with mongolism would medically justify a therapeutic abortion in order to prevent the birth of an individual who would be plagued with serious mental and physical handicaps. Before the therapeutic abortion is performed more than one doctor is usually required to testify to the medical need for the abortion. Parents should be aware of the fact that the obstetrician's religious background may affect his decision in recommending a therapeutic abortion, and should plan accordingly. Some doctors who are strict believers in certain religions, for example, the Catholic faith, may not recommend an abortion.

What Questions Might Not Be Answered By Genetic Counseling?

The genetic counselor's main concern is with helping couples to prevent passing on hereditary disorders to future children. He, in many instances, cannot give precise reasons why previous defective children were born. Many environmental as well as hereditary factors from both parents are involved in the birth of any child, and the job of a genetic counselor does not include trying "to blame" either parent for a defective child. A counselor can tell through use of amniocentesis that a future child will not have mongolism, but he cannot say that the child will be normal in all other ways; again because so many factors are involved in the formation of every human being.

What Kind of Referrals, if Any, Are Necessary for Genetic Counseling?

A person or couple may come to a genetics unit without being referred by a doctor. The genetic counselor would probably want to contact the family doctor or obstetrician, however, to gain information about the family's medical history.

How Costly Are These Tests and Genetic Counseling Procedures?

There are many genetic units which are supported by the National Foundation—March of Dimes or other organziations and offer their services and tests free of charge. Some units will analyze the amniotic fluid for free, but the patient must pay a small obstetrician's fee of about $25 for the doctor who performs the amniocentesis. A blood test and chromosome analysis for someone who has to pay the full charge will cost about $75.

Where Are Genetic Counseling Services Provided?

A couple's family doctor or obstetrician will probably be familiar with where the couple can get genetic counseling services. There are over 250 genetic units in the United States and the number of units is rapidly increasing. Some of the genetic units specialize in specific genetic diseases.

Parents may phone their nearest children's hospital or medical school to find out where chromosome tests and counseling are offered. Any local office of the National Foundation—March of Dimes can also provide information to parents or they can write to the national headquarters at P.O. Box 2000, White Plains, New York 10602.

The National Genetic Foundation also serves as a clearing house for referrals of physicians and patients and can be reached through its central telephone (212) 265-3166 which is located in New York City.

CHAPTER FIVE

GENETIC COUNSELING CAN BE APPLIED TO THE PREVENTION OF MANY OTHER BIRTH DEFECTS IN ADDITION TO MONGOLISM.

Mongolism represents only one of the more than 1,700 genetic diseases which to date have been identified and labeled.[1] Some of these defects are dominant and can be passed on if either parent is a carrier; others are recessive and can be passed on only if both parents are carriers; and still others are sex-linked and may be passed on by females and show up only in males.

Mongolism is the most common chromosome abnormal-

ity, but many different types of chromosome aberrations including double-Y, Kleinfelter's and Turner's syndromes are also found in man. Estimates have been made stating that about one child in every two hundred has a chromosome abnormality. All of the major chromosomal abnormalities are now detectable with the use of amniocentesis.

Chromosome abnormalities represent gross genetic imbalances and very productive treatment or cure is not likely. Many other defects some of which are identifiable through specimens of blood or urine can, however, be treated beneficially with special diets, drugs or therapy. Early diagnosis is important because it can often mean prevention or treatment that will allow an infant to grow up to live an almost normal life rather than being severely retarded or impaired.

What Are Some Examples of Defects That Can Be Helped by Genetics and Medical Progress?

1—A mass screening program has been started for the detection and prevention of Tay-Sachs disease, a recessive defect. Estimates have been made that 1 out of 30 American Jews of Ashkenazi descent (where approximately 90 per cent of the U.S. Jewish population originated) are Tay-Sachs carriers.[2] If both spouses are carriers the odds are 1 in 4 of having an afflicted child, and other children could be carriers.

2—A mass screening program is now available for detecting sickle cell disease which exists in all races but is particularly prevalent among Blacks.

3—The federal government approved in 1969 a vaccine that is over 99 per cent effective in the prevention of Erythroblastosis fetalis (Rh blood disease). The vaccine prevents the mother from developing the blood antibody that could seriously harm her babies.

4—A genetic counselor can tell the sister of a man with hemophilia whether or not she is a carrier of this defect.

5—Huntington's chorea is a dominant defect which does not strike and is not detectable until later in life. Children of a parent with Huntington's chorea have a 50 per cent chance of being afflicted with this disease. Because of this, adoption may be considered advisable for someone who has a parent with Huntington's chorea.

6—Cataracts which may cause blindness to a developing baby due to exposure of the mother to German measles (Rubella) can be detected through amniocentesis.

What Hereditary Birth Defects Can Be Identified Through the Use of Amniocentesis?

Amniocentesis which allows the study of fetal cells and amniotic fluid can detect prenatally many other disorders in addition to Down's syndrome. Many of the disorders are associated with severe mental retardation. The following is a partial listing of specific disorders for which identification and diagnostic procedures are currently available.[3]

1—Acatalasemia
2—Adrenogential Syndrome
3—Argininosuccinic Aciduria
4—Cataracts
5—Chromosome Abnormalities (Down's Syndrome, Turner's Syndrome,XYY, etc.)
6—Citrullinemia
7—Congenital Erythropoietic Porphyria
8—Cystathioninuria
9—Cystic Fibrosis
10—Cystinosis
11—Cytongenetic Studies
12—Erythroblastosis Fetalis (Rh blood disease)
13—Fabry's Disease
14—Fucosidosis
15—Galactosemia
16—Gaucher's Disease (Infantile & Adult Types)
17—Generalized Gangliosidosis
18—Juvenile GM1 Gangliosidosis
19—Juvenile GM2 Gangliosidosis
20—Glucose-6-PO4 Deficiency
21—Glycogen-Storage Disease
22—Histidinemia
23—Homocystinuria
24—Hunter's Disease
25—Hurler Syndrome
26—Hyperammonemia
27—Hyperlysinemia
28—Hypervalinemia
29—I-Cell Disease
30—Isovaleric Acidemia
31—Ketotic Hyperglycinemia
32—Lesch-Nyhan Syndrome
33—Lysosomal Acid Phosophatase
34—Mannosidosis
35—Maple Syrup Disease
36—Metachromatic Leukodystrophy (Late Infantile, Juvenile, and Adult Types)
37—Methylmalonic Aciduria
38—Niemann-Pick Disease
39—Ornithine-Alpha-Ketoacid Transaminase Deficiency
40—Orotic Aciduria
41—Pompe's Disease
42—Pyruvate Decarbocylase
43—Refsum's Disease
44—Sandhoff's Disease
45—San Filippo Syndrome
46—Scheie Syndrome
47—Tay-Sachs Disease
48—Virologic Studies
49—Wolman's Disease
50—Xeroderma Pigmentosum

What Are the Chances That Parents With One Severely Retarded Child Will Produce Another Similarly Affected Child?

A couple who already has one mentally retarded child has an increased risk of producing additional children with mental handicaps. The risk will depend upon the cause of retardation. Where the cause of retardation is unknown the probability of producing another severely retarded child is estimated at about 3 per cent.[4] This figures is about three times the ordinary risk.

CHAPTER SIX

THE FUTURE HOLDS THE HOPE TO THE FURTHER PREVENTION OF BIRTH DEFECTS.

Medical researchers are constantly and diligiently looking for preventive measures, treatments, and cures for birth defects. Because of this, the future holds great promise for the detection, correction, and elimination of many defects. New

advances being made each day will gradually become utilized for the welfare of the human populace.

Medical research is being prompted by such activity as the recent Congressional testimony of Nobel laureate Joshua Lederberg who estimated that 25 per cent of the nation's medical problems could now be traced in part to genetic factors.[1] Genetic diseases are, in fact, now deemed responsible for about one childhood death in five.[2]

Hereditary diseases will inevitably persist in spite of preventive medical techniques, however, because of the perpetuation of nondetectable gene carriers and the appearance of new mutations. Estimates have been made that every human being carries between five and ten potentially harmful recessive genes.[3] The immediate goal is to prevent individual cases of birth defects and, by doing so, spare sadness and grief for families who may have experienced previous misfortune due to a hereditary disorder.

What New Progress Is Being Made?

Virginia Apgar, M.D., vice-president for medical affairs of the National Foundation — March of Dimes has speculated that doctors will soon need only enough time to build an accurate family medical history and to draw blood, urine, and tissue samples in order to provide good genetic counseling.[4] The suggestion has been made by others that prenatal genetic screening through amniocentesis be made a routine part of prenatal care. The National Foundation — March of Dimes predicts that prenatal blood testing, now obligatory to determine the presence of syphilis, will be extended, especially to Rh and ABO blood groups, and to "the carrier state of some enzyme errors."[5] The goal is to detect and eliminate, or treat diseases and malformations before birth or as soon after birth as possible.

Just recently, Dr. Carl R. Merrill of the National Institute of Mental Health, as head of a research team, made the discovery that bacterial viruses may be used in the treatment of a number of genetic or hereditary diseases. Galactosemia, caused by the lack of a specific gene containing an enzyme required to metabolize sugar (galactose), may cause mental retardation, impair body functions, and even cause death. Dr. Merrill's research team found that viruses containing the bacterial gene necessary for the metabolism of galactose may be introduced into and accepted by human cells lacking this gene. This led the scientists to believe that "the 'message' contained in a specific bacterial gene can be conveyed into a functional protein by the human cells."[6] The implication is that specific

inborn errors of metabloism may be correctable in the future.

The World Health Organization which reports that at least 4 per cent of all live-born infants are suffering from genetic or partly genetic conditions is promoting genetic counseling centers throughout the world and suggests that an international register of human chromosome abnormalities be available for physicians.[7] This organization also recommends that genetic counseling services and laboratory costs be covered by health insurance.

Vaccines have been made and are available for the prevention of Rh blood disease and German measles. A mass screening program is also now in progress to detect PKU disease in newborns.

With reference to Down's syndrome, the author suggests that performing chromosome analysis on all mongoloids become standard practice. The resulting information should be forwarded to the parents so that they will be aware of risks to future children. In this way the prevalence of mongolism because of hereditary factors could be substantially reduced.

Genetic counseling centers are fast increasing in number and at least three-quarters of the more than 100 March of Dimes Birth Defects Centers across the nation offer counseling or referral to genetic services. Also increasing in number are organizations such as family counseling centers or Planned Parenthood Leagues which provide related aid.

The fears about population overcrowding make the matter of eliminating birth defects especially important. Society looks for people who will contribute to the progress of mankind, not for people who will be dependent on others and require much special care. Eliminating birth defects becomes crucial when we consider that Dr. Philip Handler, president of the National Academy of Sciences, has suggested that it is time to consider a national policy for destroying genetically unfit babies before birth.[8] In justification of this he mentioned that population growth and advancing medical skills now keep many once doomed people with genetic disorders alive long enough to reproduce themselves, and that this poses "a serious threat of damage to the human gene pool."

As has already been stated therapeutic abortions could prevent babies with serious defects from being born and leave the parents with the opportunity to try again for a normal child. Dr. Kurt Hirschhorn, president of the American Society for Human Genetics, has calculated that 14 million abortions over the next 40 years could completely eliminate cystic fibrosis and sickle cell anemia, two of the more common inherited diseases, and help prevent other abnormalities.[9]

The future waits for more advances to be made and the public waits to utilize these new developing techniques. Educating the public of services available to them is now the present goal for the future.

Who Is Responsible?

Members of the medical profession and related social service agencies are responsible for disseminating information on medical advances that are ready to be utilized by the public. The public, on the other hand, is responsible for being mature enough to be informed and to make use of services available to them.

As will be made increasingly clear the right to be born is becoming qualified by "the right to have a reasonable chance of a happy and useful life." It is likely that the future may see controversial law suits being brought against parents for producing defective children through neglect or refusal to use preventive measures available. Some "wrongful life suits" have already been made on behalf of victims of infectious syphilis and German measles.[10] The future is in the hands of those who will make something of it by using all the resources available to them. The author hopes that you do.

GLOSSARY OF COMMON TERMS

aberration—A change from the normal.
abortion—The removal of the fetus (developing baby) from a pregnant woman.
abnormality—Different from the normal, usually in a negative way.
afflicted—Having a condition which is harmful and limiting.
amniocentesis—The process by which a sample of amniotic fluid is obtained from a pregnant woman.
amniotic fluid—The fluid surrounding the developing baby.
birth defects—Defects or abnormalities present at birth which may be apparent then or show up at a later time. The causes of birth defects may be hereditary, environmental, a combination of these factors, or unknown.
carrier—A person who may pass on a hereditary birth defect. This person is not necessarily afflicted with the defect, himself.
chromosomes—Units of heredity which carry the genes. The chromosomes usually come in pairs, one of each pair being donated from each parent.
chromosome analysis—A process by which chromosomes are studied to determine that they are correct in number and arrangement.
congenital—Present at birth, regardless of whether it is apparent at that time.
defect—A negative difference from the normal which will usually lead to either a mental or physical handicap, or both.
fetus—A developing baby in a pregnant woman.
gene—The smaller units of heredity carried by the chromosomes. The genes are responsible for determining a person's general physical and mental qualities.
genetics—The study of the genes as agents of heredity.
geneticists—Scientists studying genetics.
genetic counseling—Counseling individuals about procedures available for detecting and diagnosing hereditary defects; predicting the probability of the defects reoccurring; detecting carriers of the defects; preventing the defects; and eliminating the defects. Genetic counseling need not include specific recommendations of actions to be followed, but should include information on procedures which could be useful in each particular case.
hereditary—Some physical or mental trait that has been or can be passed on to children by their parents.
inborn error—A biochemical disorder related to the genes; one of which may be missing or faulty, and not producing a specifically needed enzyme (a protein which acts as a catalyst for chemical reactions in the body).
intelligence—The mental capabilities of an individual.
intrauterine diagnosis—The examination of a woman's uterus.
karyotype—A greatly enlarged picture of the chromosomes of an individual showing the chromosome numbers and arrangement.
metabolism—The processes involving the chemical reactions of the body.
mongolism—A birth defect caused by an extra #21 chromosome or equivalent #21 chromosome material translocated to another chromosome. This defect usually causes moderate to severe retardation to the afflicted individual and is hereditary in some cases.
mutation—A genetic change which can be transmitted to future

children. Mutations may be caused by exposure to harmful environmental agents or be genetic accidents for which there is presently no answer.

nondisjunction—The failure of a chromosome pair to separate during cell division.

pedigree—A chart of a family's medical history.

sibling—A brother or sister of a child borne of the same parent.

therapeutic abortion—An abortion performed for medical reasons in the interests of a developing baby or the pregnant woman, or both.

translocation—A chromosome is translocated when it is attached to another chromosome and carried by that chromosome.

trisomy—A trisomy of a chromosome is an extra and separate chromosome in addition to the normal chromosome pair.

uterus—The place in a woman where the baby develops.

NOTES

The following notes are given for those seeking further information on points that have been made in each chapter or seeking sources for information that has been reported.

CHAPTER ONE

1. **Facts on Mental Retardation.** Texas: National Association for Retarded Children, 1971, p. 5.
2. **With Best Wishes For A Happy Birth Day,** New York: The National Foundation—March of Dimes, p. 1.
3. Theodore Friedmann, "Prenatal Diagnosis of Genetic Disease," **Scientific American,** (Nov., 1971), p. 38.
4. United States Department of Health, Education, and Welfare, **Mongolism (Down's Syndrome),** Public Health Service Publication #720, Health Information Series #94, Washington: U. S. Government Printing Office, 1970, p. 3.
5. **Chromosome 21 and its association with Down's Syndrome,** New York: The National Foundation—March of Dimes.
6. **Mongolism (Down's Syndrome),** p. 3.

CHAPTER TWO

1. James S. Thompson, M. D. and Margaret W. Thompson, Ph.D., **Genetics in Medicine,** Philadelphia: W. B. Saunders Company, 1966, p. 104.
2. " 'Clinical genetics' gain notice," **Medical News,** Vol. 213, No. 13 (September 28, 1970), p. 2171.
3. Cyrus W. Stinson, M. D., **Understanding the Mongoloid Child,** Texas: National Association for Retarded Children, 1971, p. 4.
4. Thompson and Thompson, p. 105.

CHAPTER THREE

1. "Genetic Counseling Unveils Secrets of Unborn," The **Health Bulletin,** (June, 1971).
2. Daniel Bergsma, M.D. (ed), **Intrauterine Diagnosis,** VII, No. 5, New York: National Foundation—March of Dimes, 1971, p. 5.
3. This table was adapted from a table "Criteria For Doing Amniocentesis" in the Daniel Bergsma and Henry T. Lynch, eds., **International Directory of Genetic Services,** New York: National Foundation —March of Dimes, 1971, pp. 49-50.
4. **Intrauterine Diagnosis,** p. 8.

CHAPTER FOUR

1. United States Department of Health, Education, and Welfare, **Mental Retardation, its biological factors,** Public Health Service Publication #1152, Health Information Series #114, Washington: U. S. Government Printing Office, 1970, p. 16.

CHAPTER FIVE

1. " 'Clinical genetics' gain notice," **Medical News,** Vol. 213, No. 13 (Sepetmber 28, 1970), p. 2168.
2. "Mass tests begin to find Tay-Sachs gene carriers," **Medical News,** Vol. 216, No. 7 (May 17, 1971), p. 1097.
3. This table was adapted from the **International Directory of Genetic Services,** p. 44 and **Scientific American,** p. 42.

4. Brian Kirman, M.D., "Genetic Counseling for parents of mentally handicapped children," **Mental Retardation,** Canadian ARC, Vol. 71, No. 2 (April, 1971), p. 12-13.

CHAPTER SIX

1. " 'Clinical genetics' gain notice," **Medical News,** Vol. 213, No. 13 (September 28, 1970), p. 2167.

2. **Genetic Counseling and Treatment; A Major Challenge for Medicine,** National Foundation for Genetics and Neuromuscular Diseases, Inc. (February 4, 1970), p. 1.

3. **Ibid.,** p. 9.

4. " 'Clinical genetics' gain notice," **Medical News,** p. 2169.

5. **With Best Wishes For a Happy Birth Day,** New York: The National Foundation—March of Dimes, p. 9.

6. Bertram S. Brown, M.D., "Viruses for M.R. Cure?," **Montachusett (Mass.) Review,** Vol. 8, No. 31 (March 8, 1972), pp. 24-25.

7. "Genetic Counseling: Third Report," World Health Organization Technical Report Series No. 416, (Geneva Switzerland, 1969).

8. "Aborting Unfit Babies in Fetus Viewed," **Boston Record American,** October 23, 1971, p. 4.

9. "Intelligence Report," **Boston Sunday Globe,** September 21, 1969, p. 4.

10. Theodore Friedmann, "Prenatal Diagnosis of Genetic Disease," **Scientific American,** (Nov., 1971), p. 42.